Management of femoral fractures in adult onset osteopetrosis: A multicentric retrospective study

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Abstract

Background: Osteopetrosis is a relatively rare disease with an incidence of approximately 1 case per 2,00,000 to 5,00,000 population. The clinical features of osteopetrosis include pathological fractures, severe anemia due to defective erythropoiesis, bone marrow failure, coagulation defects, propensity for developing severe infections. Common fractures seen in these patients include hip and femoral fractures. Management of fractures in these patients is technically difficult and postoperative complications are common. We conducted a retrospective study of 20 patients with osteopetrosis who presented with femoral fractures and were treated in our institute.

Materials and Methods: This was a multicentric retrospective study of 20 patients with osteopetrosis who presented with femoral fractures. The patients were included in this study on the basis of predefined inclusion and exclusion criteria. The diagnosis was based upon imaging studies (X-Rays in all the patients and CT in selected cases). The operative procedure post-operative complications, functional outcome and complications during follow up were studied from case papers. The data was tabulated and analyzed using Minitab 17 version software.

Results: Out of 20 studied cases there were 16 (80%) males and 4 (20%) females with a M:F ratio of 1:0.25. The most common affected age group was found to be between 30 – 35 years (35%). Majority of patient sustained fracture following motor vehicular accidents (65%) followed by fall from height (20%) and direct blow or assault (15%). 13 patients were found to be having co-morbidities including anemia, carpal tunnel syndrome and visual disturbances. Open reduction and internal fixation was done in all the cases. Technical difficulties seen during operative procedures included difficult because of obliterated canal, sclerosed medulla and dense swarf. Common complications in these patients included wound infection, hematoma formation and arthritis of hip joint (5%). Repeat surgeries were required in 3 (15%) patients and 1 (5%) patient developed non-union of the fracture ends.

Conclusion: Management of femoral fracture in patients with osteopetrosis is challenging for operating orthopedic surgeon. Common problems encountered during open reduction and internal fixation include overheating during drilling, smoke production and increased temperature of both bone and drill bits and production of dense bone swarf. Orthopedic surgeon should be aware of these technical difficulties encountered during operative interventions.

Keywords: Osteopetrosis, femoral fractures, open reduction and internal fixation, drilling

Introduction

Osteopetrosis is a relatively rare disease with an incidence of approximately 1 case per 2,00,000 to 5,00,000 population \(^1\). The basic pathology in osteopetrosis is failure of bone resorption by osteoclasts due to which there is defective bone modelling and remodeling. This defective bone remodeling leads to skeletal fragility despite an apparent increase in bone mass. The secondary consequences of such an increase in bone mass, along with bone fragility, is associated with secondary complications such as pathological fractures, defective hematopoiesis, dental defects, nerve entrapments, growth disturbances and renal tubular acidosis \(^2\). Frontal bossing may be seen in pediatric age group. Osteopetrosis is divided into infantile, intermediate and adult onset forms on the basis of age of onset and clinical features \(^3\). The clinical features of osteopetrosis includes pathological fractures, severe anemia due to defective erythropoiesis, bone marrow failure, coagulation defects, propensity for developing severe infections \(^4\).
The infantile form osteoporosis usually causes growth retardation, infections and death usually occur by the 1st decade of life due to severe anemia and congestive cardiac failure. Adult onset osteoporosis may present with bony abnormalities such as marked sclerosis of skull and other bones. There is an abnormally high risk of fractures due to increased fragility of bones. Other features of adult onset osteoporosis includes cranial neuropathies due to nerve entrapments, carpal tunnel and tarsal tunnel syndrome, osteoarthritis and osteomyelitis especially of mandible. Non-skeletal manifestation may include visual impairment due to retinal detachment, retinal degeneration and psychomotor retardation. The diagnosis of osteoporosis is generally made on the basis of imaging. The imaging features are distinct in infantile as well as adult form of osteoporosis. Autosomal dominant adult type of osteoporosis usually presents with pronounced Osteosclerosis of cranial vault with clinical presentation as cranial nerve palsies (Type I) or thickening of endplate, also known as sandwich vertebra, and classical bone-within-bone appearance in the pelvis (Type II). Irrespective of the type of osteoporosis a generalized increased in the bone density is the hallmark of osteoporosis. Though not diagnostic, Hypocalcemia, hyperparathyroidism and increased levels of acid phosphatase and Creatinine kinase isof orm BB are the other biochemical abnormalities seen in these patients.

Pathological fractures are one of the hallmarks feature of the affected patients. Defective modelling and remodeling is responsible for increasing fragility of bones predisposing them for pathological fractures. Relatively low-energy or trivial fall may cause fractures in these patients. Many of the patients with osteoporosis remain asymptomatic till they present with pathological fracture. The hip and femur fractures are common pathological fractures seen in these patients. The management of these fractures are challenging because of defective bone remodeling and increased bone density. Moreover, fixation of an osteoporotic femoral fracture is difficult and is open to complications. Delayed union, malunion, non-union and Varus deformities of these fractures are some of the common complications seen in these patients. Even in otherwise uneventful open reduction and internal fixation cases these patients are predisposed for development of infections which may further complicate the management.

We conducted this retrospective study of 20 patients with osteoporosis who presented with femoral fracture and were treated in our institute.

Materials and Methods
This was a multicentric retrospective study of patients who presented with femoral fracture who were either known cases of osteoporosis or were diagnosis to be having osteoporosis for the first time after they were admitted for fracture of femur. A total of 20 patients who had been admitted for femoral fractures were diagnosed or were known to be having osteoporosis. The cases were included in the study on the basis of a predefined inclusion criteria. Cases having any of the exclusion criteria were excluded from the study. A detailed history was taken in all the patients specially to know similar complaints in any other family members, history of past fractures and other significant illness in past. History of blood transfusion was asked. Previous investigation, if available, were reviewed.

Operative Procedure
Under general anesthesia, the patient was placed into the right or left decubitus position depending upon the side of fracture and the affected extremity was prepared. Patients were operated under general anesthesia. The fracture line was opened with a longitudinal incision from the lateral side. After the fracture ends were reached the medulla was drilled. In almost all cases there was sclerotic medulla and it was difficult to drill the bone with routine drills. In many of the cases the medullary canal was obliterated. For drilling purposes, A 2.5 mm high-speed steel (HSS) drill bit was used with saline cooling. Low speed and high torque setting were used. During drilling a dense bone swarf was frequently encountered that required frequent removing of drill and saline irrigation to clear the accumulated swarf. Drilling was found to be technically very difficult because of obliterated canal, sclerosed medulla and dense swarf. The fracture was then reduced and fixation was made with screws. Bone specimens which were obtained by drilling and the intraoperative biopsy were sent for pathological examination. The histopathological examination confirmed the diagnosis of osteoporosis. Fluoroscopy checking was applied and the operation was concluded. On the radiograph taken on postoperative Day 2. If the screws were found to be in place and proper reduction was confirmed then the patients were referred for the rehabilitation program on the 4-5th postoperative day and sutures were removed at 15 days. Patients were mobilized without loading on the healing fracture, using a crutch at two weeks and with partial loading at six weeks.

Fig 1: An anteroposterior view of the right hip at the time of initial presentation shows femoral fracture and the Osteosclerotic appearance of the Osteoporotic bone (Left). Postoperative radiograph after operation with proximal femoral anatomic plate and screws (Right).

The follow up records of at least 1 year was reviewed in all the patients. The data was collected from the case papers of the patients. Statistical analysis was done with Minitab version 17 running on windows 10. Microsoft word was used for manuscript preparation while excel was used for creation of figures and graphs.

Inclusion Criteria
1. All patients who were admitted with femoral fractures and were either know case of osteoporosis or were diagnosed to be having osteoporosis after admission to our institute.
2. Age of the patients should be more than 18 years.
3. The case papers of all the patients included in this study had complete record of treatment as well as of follow up visits for at least 1 year after the discharge of patients from our institute.

Exclusion Criteria
1. Patients who have undergone failed attempts at open reduction and internal fixation prior to being referred to our institute.
2. Patients in whom treatment record was not complete or missing.
3. Follow up record for at least 1 year was not present.
4. Age less than 18 years.

Results
This was a retrospective study of patients of osteopetrosis admitted with femoral fracture. Hospital records of patients were studied for demographic details. Out of 20 patients who met the inclusion criteria of the study there were 16 (80%) males and 4 (20%) females with a M:F ratio of 1:0.25.

Majority of patient sustained fracture following motor vehicular accidents (65%) followed by fall from height (20%) and direct blow or assault (15%).

In 9 (45%) patients fracture of femur was on right side while in 7 (35%) patients was fracture was on seen on left side. Bilateral Femoral Fractures were seen in 4 (20%) patients.

The analysis of the cases on the basis of associated co-morbidities showed that out of 20 patients 7 were not having any co-morbidities. Anemia was present in 5 patients while carpal tunnel syndrome was present in 3 patients. Other co-morbidities present were osteomyelitis (10%), visual disturbance (10%) and facial nerve palsy (5%).
The classification of fractures on the basis of imaging showed that out of 20 studied cases 7 patients had simple fractures of proximal femur whereas 8 patients had wedge fractures. Remaining 5 patients had complex fractures of distal femur.

All patients were treated by open reduction and internal fixation as per the institutional protocol. Patients were operated under general anesthesia. The fracture line was opened with a longitudinal incision from the lateral side. After the fracture ends were reached the medulla was dri...
tissue or increased destruction of red cells or a combination of both these factors [19]. Anemia in adult onset osteopetrosis is difficult to diagnose as most of these patients usually remain asymptomatic and the anemia is usually detected when these patients are investigated for other purposes (preoperative evaluation in cases of fractures) [19].

The management of fractures in osteopetrosis is challenging as it is difficult to achieve proper fixation in these patients. The technical difficulties in drilling in these patients are well known to orthopedic surgeons. There are many instances of breaking of drills during drilling of bones in patients with osteopetrosis. Moreover, screw fixation is also a challenging task due to the strength required. The technical difficulties while drilling bones in patients with osteopetrosis include overheating during drilling, smoke production and increased temperature of both bone and drill bits and production of dense bone swarf. Low speed drilling, frequent changing of drill bits and continuous saline irrigation are some of the measures which can be taken to overcome these difficulties while doing drilling in these patients [20].

The outcome of management of femoral fractures in patients with osteopetrosis is variable and complications such as nonunion or varus malunion are common. In some cases, revision surgeries may also be required. Orthopedic surgeons should be aware of possible challenges during treatment of such patients.

Conclusion
The patients with osteopetrosis are prone for fractures following trivial trauma. Management of these fractures is complicated by factors such as overheating during drilling, smoke production and increased temperature of both bone and drill bits and production of dense bone swarf. Moreover, fractures in these patients may be complicated by malunion and nonunion. It is essential for operating orthopedic surgeon to be aware of intraoperative technical difficulties and postoperative complications seen in these patients.

References